These Idua-W392X mice carry a nonsense mutation of Idua (alpha-L-iduronidase) that is analogous to the W402X mutation commonly found in patients with Hurler syndrome and are valuable for evaluation of therapeutic treatments for mucopolysaccharidosis type I-Hurler (MPS I-H).

Donating Investigator
Kim M. Keeling, University of Alabama at Birmingham

**GENETIC OVERVIEW**

**Idua**<sup>tm1.1Kmke</sup>

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<thead>
<tr>
<th>Allele Type</th>
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**RESEARCH APPLICATIONS**

Developmental Biology Research
Internal/Organ Research
Details

Detailed Description

These mice carry a nonsense mutation at Idua codon W392, (Idua-W392X), which corresponds to the human IDUA-W402X mutation that prematurely terminates protein synthesis and is commonly found in patients with mucopolysaccharidosis type I-Hurler (MPS I-H) syndrome. Deficiency of the alpha-L-iduronidase enzyme causes this lysosomal storage disease. No alpha-L-iduronidase activity is detected in brain and liver tissues from homozygous mice aged 5, 10 and 30 weeks. Although mice that are homozygous for this mutation are viable and fertile, they have a median lifespan of 69 weeks. Homozygotes exhibit a progressive increase in urinary excretion of glycosaminoglycans (GAGs), and progressive accumulation of GAGs in tissues. The level of steady state Idua mRNA is reduced 30-50%. Histological analysis reveals progressive accumulation of lysosomal storage inclusions in the cytoplasm of Purkinje cells, medullar neurons, as well as infiltration of foamy macrophages that increases with age. Thickening of the zygomatic arch and femur is detected at 15 weeks of age by radiography, and increases at 35 weeks of age. At 35 weeks of age femur bone mineral density is increased and percent body fat decreased.

Development

Control Suggestions

Selected References

Genetics

Idua<sup>tm1.1Kmke</sup>

Disease/Phenotype

Disease Terms

Research Areas By Phenotype
Genotyping Protocols
Standard PCR: Idua alternate1
Probe: Idua Probe
Genotyping resources and troubleshooting

Dietary Information
LabDiet® 5K52 formulation (6% fat)

Breeding Considerations
When maintaining a live colony, these mice can be bred as homozygotes; however, homozygotes have a median lifespan of 69 weeks.

Additional Breeding and Husbandry Support
Mating System
Homozygote x Heterozygote
Heterozygote x Homozygote

Citation
When using the Idua-W392X mouse strain in a publication, please cite the originating article(s) and include JAX stock #017681 in your Materials and Methods section.

Animal Health Reports
Facility Barrier Level Descriptions


Live Mouse

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Related Products and Services

Frozen Mouse Embryo

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Breeder Pair

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All Related Strains